

| Disease | Method | Sample Size |
|--|-----------|--|
| Prader-Willi syndrome | FDNA | 31 cases in training; 17 cases in testing |
| Prader-Willi syndrome | Face2Gene | 49 cases |
| Unilateral hypohidrotic ectodermal dysplasia | Face2Gene | 136 cases, 717 con |
| Edward syndrome (ES) Klinefelter syndrome (PKS) | Face2Gene | 59 ES, 70 PKS, 973 cc 973 others |
| Reye-Gripp syndrome | Face2Gene | 13 cases, 20 controls, |
| Cholesterolosis type IV | Face2Gene | 26 cases, 98 controls, 9 |
| Albinism | Face2Gene | 14 cases |
| Congenital adrenal hyperplasia | DNN | 102 cases, 144 con |
| Down syndrome (NS) Williams syndrome (WBS) | LBP, SVM | 286 NS, 161 WE |

Abbreviations and explanations: cases, patients; controls, patients without genetic disorders; FDNA, facial dysmorphology novel analysis; Face2Gene, Face2Gene; AUC, area under the curve; DS, Down syndrome; DNN, Deep Neural Network; LBP, Linear Binary Probability; SVM, Support Vector Machines.

Diagnostic Recognition Of Genetic Disease

SA Dillow



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